

Pedigree Studies

25

Pedigrees are not reserved for show dogs and race horses. All living things, including humans, have pedigrees. A pedigree is a diagram that shows the occurrence and appearance, or phenotype, of a particular genetic trait from one generation to the next in a family. Genotypes for individuals in a pedigree usually can be determined with an understanding of inheritance and probability.

In this investigation, you will

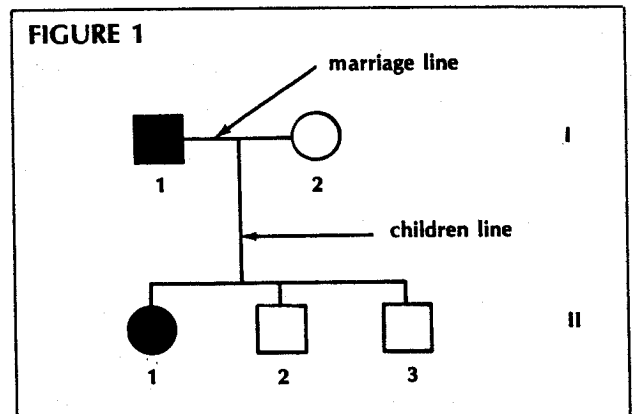
- learn the meaning of all symbols and lines that are used in a pedigree.
- calculate expected genotypes for all individuals shown in pedigrees.

Procedure

Part A. Background Information

The pedigree in Figure 1 shows the pattern of inheritance in a family for a specific trait. The trait being shown is earlobe shape. Geneticists recognize two general earlobe shapes, free lobes and attached lobes (Figure 2). The gene responsible for free lobes (*E*) is dominant over the gene for attached lobes (*e*).

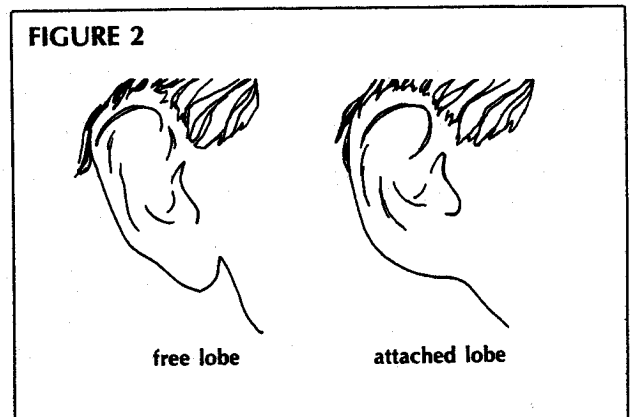
In a pedigree, each generation is represented by a Roman numeral. Each person in a generation is numbered. Thus each person can be identified by a generation numeral and individual number. Males are represented by squares whereas females are represented by circles.



Part B. Reading a Pedigree

In Figure 1, persons I-1 and I-2 are the parents. The line which connects them is called a marriage line. Persons II-1, 2, and 3 are their children. The line which extends down from the marriage line is the children line. The children are placed left to right in order of their births. That is, the oldest child is always on the left.

- What sex is the oldest child? _____
- What sex is the youngest child? _____



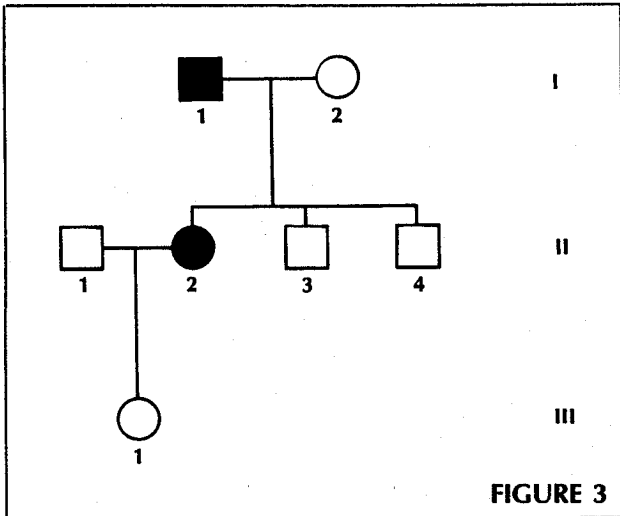


FIGURE 3

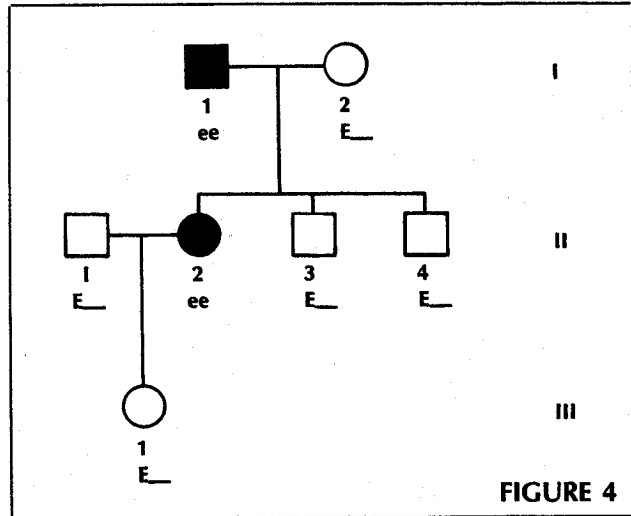


FIGURE 4

Using a different pedigree of the same family at a later time shows three generations. Figure 3 shows a son-in-law as well as a grandchild. Generation I may now be called grandparents.

3. Which person is the son-in-law? _____
4. To whom is he married? _____
5. What sex is their child? _____

Part C. Determining Genotypes from a Pedigree

The value of a pedigree is that it can help predict the genes (genotype) of each person for a certain trait.

All shaded symbols on a pedigree represent individuals who are homozygous recessive for the trait being studied. Therefore, persons I-1 and II-2 have ee genotypes. They are the only two individuals who are homozygous recessive and show the recessive trait. They have attached earlobes.

All unshaded symbols represent individuals who have at least one dominant gene. These persons show the dominant trait.

To predict the genotypes for each person in a pedigree, there are two rules you must follow.

Rule 1. Assign two recessive genes to any person on a pedigree whose symbol is shaded. (These persons show the recessive trait being studied.) Small letters are written below the person's symbol.

Rule 2. Assign one dominant gene to any person on a pedigree whose symbol is unshaded. (These persons show the dominant trait being studied.) A capital letter is written below the person's symbol.

These two rules allow one to predict some of the genes for the persons in a pedigree. Figure 4 shows the genes predicted by using these two rules.

To determine the second gene for persons who show the dominant trait, a Punnett square is used. In Figure 4, we already know that the grandfather (I-1) is ee . If the grandmother (I-2) were EE , could any ee children (like II-2) be produced? A Punnett square shows this combination to be impossible. Thus, the grandmother must be heterozygous or Ee .

6. (a) Can an Ee parent and an ee parent have the results shown in generation II? _____
- (b) Prove your answer by showing the results in the below left Punnett square.

	e	e
E		
e		

	E	E
e		
e		

- (c) Can an EE parent and an ee parent have the results shown in Generation II? _____
- (d) Prove your answer to the above question by showing the results in the above right Punnett Square.

7. (a) Predict the second gene for person II-3.
(Read it from the Punnett square.) _____

- (b) Predict the second gene for person II-4.

(c) Could child II-3 or II-4 be *EE*? _____

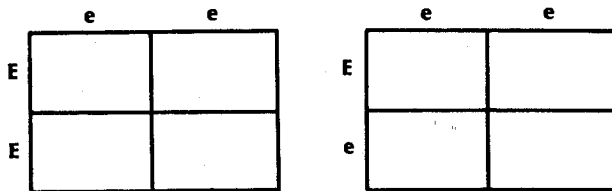
Explain. _____

To predict the second gene for person II-1, a different method must be used, since he could be either *EE* or *Ee*.

8. (a) Can an *EE* person married to an *ee* person (II-2) have children with free earlobes?

(b) Can an *Ee* person married to an *ee* person

have children with free earlobes? _____
 (c) Prove your answers by showing the results of these crosses in the Punnett squares below.



In this case, the second gene from person II-1 cannot be predicted using Punnett squares. Either genotype *Ee* or *EE* may be correct. When this situation occurs, both genotypes are written under the symbol (Figure 5).

Predicting the second gene for III-1 results in her being heterozygous. Although her mother must provide her with one recessive gene, she has free lobes, so the second gene must be dominant (Figure 5).

At some time in the future, if II-1 and II-2 have many more children, one might be able to predict the father's second gene. For example, if they have ten children and all show the dominant free lobes, one could safely conclude that he is *EE*. If, however, they have some children with attached earlobes (*ee*), then he must be *Ee*.

Analysis

1. Draw a pedigree for a family showing two parents and four children.

- (a) include a marriage line and label it.
- (b) include a children's line and label it.
- (c) make the oldest two children boys and the youngest two girls.

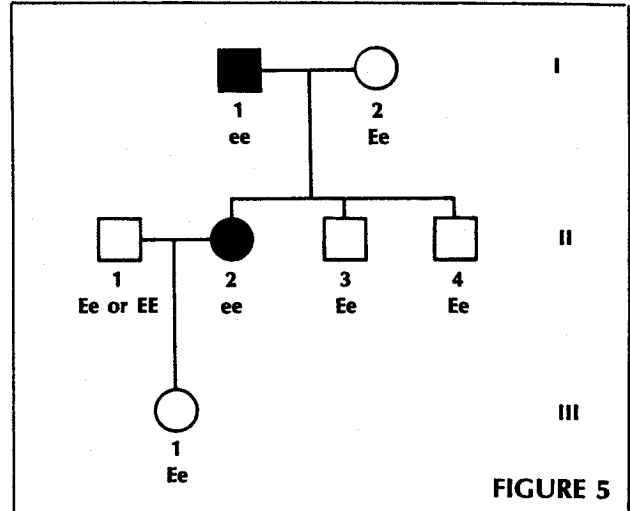
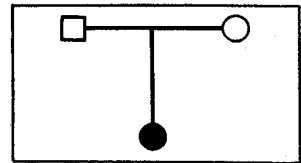


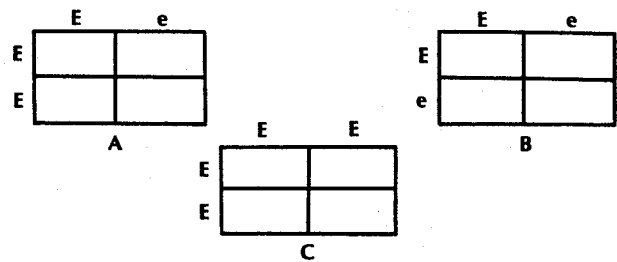
FIGURE 5

When both parents show a dominant trait and their child or children all show a dominant trait, one cannot predict the second gene for anyone if only a small family is available.

Examine this pedigree:

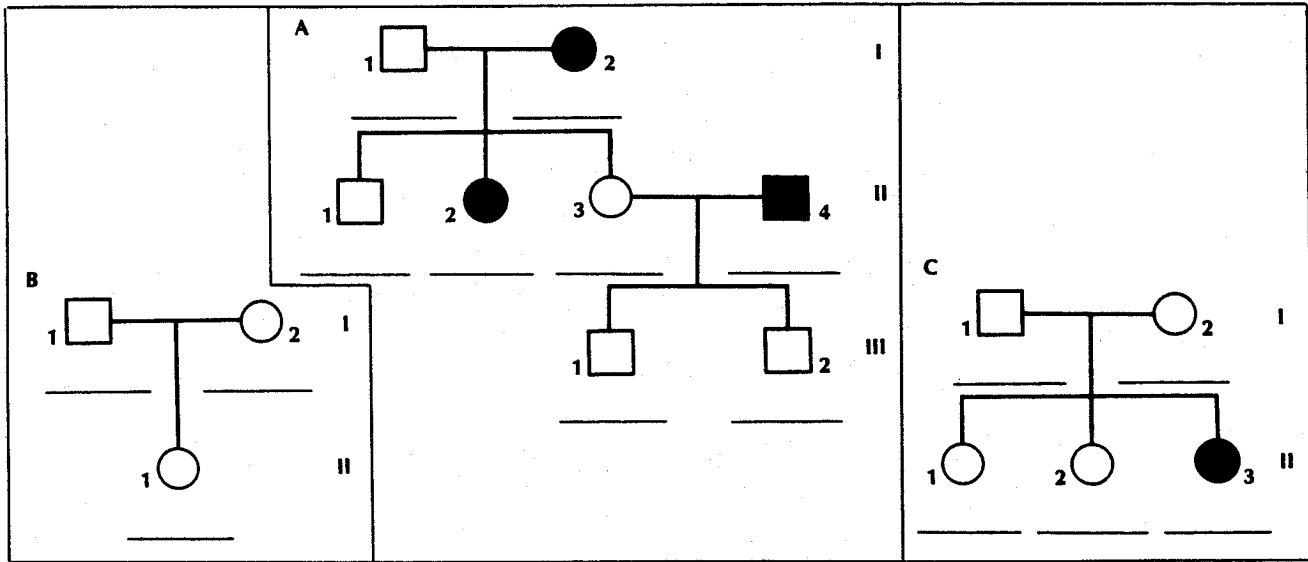


9. (a) Which Punnett square, A, B, or C, would best fit this family? _____

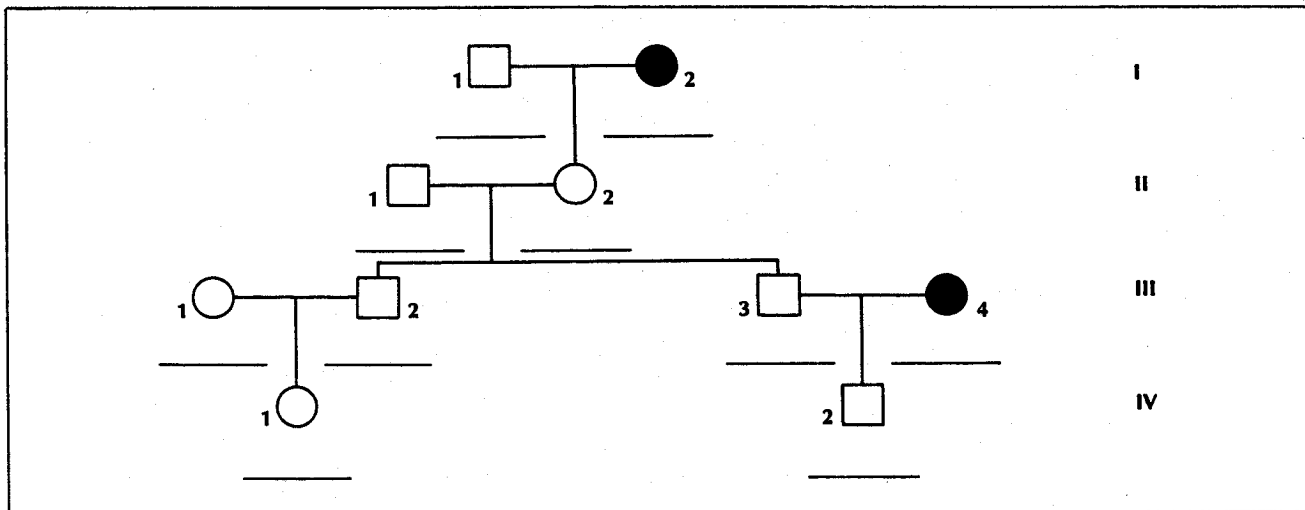


(b) Explain. _____

- Using the pedigree from question 1, indicate that person II-2 has attached earlobes.
- Using the pedigrees below, predict the genotypes for these families. (Remember the two rules—first give all shaded symbols two recessive genes and give unshaded symbols one dominant gene.) Write the letters on the lines provided.



- Examine the pedigree below.



- How many generations are shown? _____
- How many persons have free earlobes? _____
- How many persons have attached earlobes? _____
- Identify by generation and number those persons with attached earlobes. _____

- Give the genotype for all persons having attached earlobes. _____
- How many children did the original generation have? _____

- Predict the genotypes for all persons in question 4 using the lines below each person's symbol.